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Who and how we engage: A systemic mapping of stakeholder perspectives on genomic newborn screening

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The use of genomic sequencing in newborn screening is increasingly being piloted through research programmes worldwide. In parallel, an expanding body of attitudinal studies has examined stakeholder views as jurisdictions consider whether and how to implement genomic newborn screening (gNBS). To understand what perspectives have been represented thus far in stakeholder research and how those perspectives have been shaped by study design, we conducted a systemic mapping of 41 attitudinal studies about gNBS. Studies were conducted in eight countries, with more than half linked to ongoing pilot programmes. We observed a limited range of perspectives, with mothers and people with prior experience of a genetic condition in the family relatively frequently represented. As gNBS would primarily involve testing newborns who are not affected by genetic conditions, additional perspectives are needed. Studies varied considerably in how gNBS was described to participants. The issues most frequently explored concerned acceptability ($n = 31$), consent ($n = 24$), scope of screening ($n = 26$), psychosocial implications ($n = 25$), and data storage and secondary use ($n = 22$), whereas healthcare system readiness ($n = 13$) and equity considerations ($n = 11$) received comparatively less attention. Many studies left unaddressed participants' underlying assumptions about the predictive certainty of genomic findings, highlighting a need to situate gNBS within the realities of genomic uncertainty. At a time when policymakers are actively considering gNBS, this mapping highlights the need for careful interpretation of existing findings and identifies research priorities, particularly in relation to understanding gNBS as a public health programme.

European Journal of Human Genetics; https://doi.org/10.1038/s41431-026-02185-9

INTRODUCTION

Since population-based newborn screening (NBS) began in the 1960s, it has become one of the most successful public health initiatives, producing significant individual and population-level benefits by reducing mortality and morbidity [1, 2]. Decisions about which conditions to include in NBS have to date been made on a condition-by-condition basis, weighing the potential benefits and harms of screening [3].

Technological advances mean that genomic sequencing could radically expand the scope of NBS (gNBS), enabling the detection of thousands of genetic conditions not identifiable through existing methods. More than thirty pilot studies worldwide have already been launched [4], and to date, over 352,000 babies have been enroled [5]. Most of these prioritise screening for highly-penetrant treatable conditions, yet understanding of severity, penetrance, and expressivity from gNBS findings is a result of significant ascertainment bias - meaning that findings do not always predict how or if an individual will be affected. However, this nuance is often lost amid prevailing narratives of genetic determinism that risk fostering unrealistic expectations of what gNBS can deliver.

The expansion of newborn screening through the use of genomic sequencing prompts debates around which genomic variants provide useful predictions and should therefore be reported to parents; how to balance uncertainty with early detection and how genomic data should be stored, governed, and potentially reinterpreted over time. To help inform gNBS studies and emerging policies and examine the ethical issues raised by this technological shift, it is critical that the views and experiences of stakeholders are taken into account. Several reviews have already synthesised existing research on attitudes towards gNBS [6–8], highlighting perceived benefits and challenges. The purpose of this article therefore is not to consolidate these perspectives, but rather to critically examine how – and from whom – such perspectives have been generated. Analysis of stakeholder views risks being incomplete without a critical examination of the methods used to elicit them. We therefore analyse who has been included in gNBS opinion studies, how issues have been framed, and what perspectives may still be missing (e.g. questions that have not yet been explored, or stakeholders who have not yet been involved). As policymakers and advisory bodies are actively seeking evidence on stakeholder attitudes to guide decisions about gNBS, this work to identify gaps for further research is especially timely.

METHODS

Search strategy

We used several strategies to gather relevant documents. First, we performed searches of MEDLINE, Embase, International Bibliography of the Social Sciences (IBSS), Web of Science, PsycINFO, PROSPERO, Social

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Received: 31 January 2026 Revised: 23 June 2026 Accepted: 30 June 2026
Published online: 20 July 2026

Sciences Citation Index (SSCI), TRIP, and Google Scholar. Searches were conducted (by DK under the supervision of AP and LJ) in June 2024 using combinations of keywords relating to study design, sequencing technology, and newborn screening. An example search string was:

(qualitative OR "focus group" OR interview OR survey OR questionnaire OR perspective OR attitude)

AND

("whole genome sequencing" OR "whole exome sequencing" OR "WGS" OR "sequence analysis")

AND

("newborn screening" OR "neonatal screening")

Second, we searched websites of members of the International Consortium on Newborn Sequencing (ICoNS) and supplemented this search with relevant organisations such as national screening committee pages.

Third, we supplemented our search via snowballing.

Eligibility criteria

All references were imported into Covidence, which was used for duplicate removal and screening.

We included empirical studies and reports on stakeholder perspectives towards gNBS. Eligible publications reported empirical data (qualitative, quantitative, and mixed-methods research) or reported on patient and public engagement activities. Protocols were not included in the primary analysis. We excluded reviews, commentaries, purely normative or conceptual articles, and studies not in English.

274 references were imported from our initial search and following the removal of duplicates and screening of titles, abstracts, and full articles, 41 studies met the inclusion criteria (see Fig. 1). Articles where eligibility was unclear were discussed with an additional senior member of the research team.

Data extraction and analysis

Data were collated in a structured spreadsheet and analysed descriptively to map the scope, methods, and findings of the included studies. More specifically, extracted information included: study details (authors, affiliated research group, year, country, location), participant characteristics and sample size, recruitment methods, data collection methods, participant briefing methods, main themes, and outcomes.

Data extraction was piloted independently by two researchers (AP and LJ) to ensure consistency. Following this, one researcher (AP) extracted data from all included studies, with uncertainties discussed within the team (AP and LJ).

RESULTS

41 of documents met the inclusion criteria (see Table 1 for full report of included documents) [9–48]. In addition, we found 5 protocols [49–53]. The countries represented were the USA, UK, Australia, Canada, New Zealand, Slovenia, Bulgaria, and China. Documents were published between 2014 and 2024.

Our analysis of current gNBS engagement approaches included the following categories: who are the stakeholders and how are the gNBS issues constructed (Fig. 2).

Who were the stakeholders?

A wide range of participant groups were represented across the included studies, with some studies engaging mixed samples. Four main categories of stakeholders emerged: parents, the public, healthcare professionals, and other experts.

Parents (both current and prospective) were the most frequently sampled group. Across studies, this group was heterogeneous, encompassing parents of newborns who had recently undergone screening, parents of children with genetic conditions or prior testing experiences, and expectant parents or parents from the general population without direct experience of newborn screening. Mothers were more represented than fathers, reflecting both study design choices (e.g., recruitment through maternity clinics) and broader participation trends. This imbalance resulted in an underrepresentation of paternal perspectives. The public was also frequently engaged, though the distinction between "parents" and "publics" was often blurred. Many participants described as members of the public had personal or family experience with genetic testing or rare disease [12–15, 19–21, 27, 32, 39, 41–44, 47]. In some cases, these experiences were highly prevalent within samples, raising questions about whether findings reported as "public perspectives" instead largely reflect the views of the rare disease community. To interpret findings, it may therefore be more useful to distinguish between participants with and without personal or familial experience of genetic disease, rather than between "parents" and the "public." Public samples ranged in age from children [44] to older adults [42, 43], and recruitment methods included social media [41–43], advocacy groups [30, 35, 41, 44], clinics [12, 13, 15, 17, 18, 22, 23, 25, 32, 37–39], and market research companies [9, 19, 21, 24].

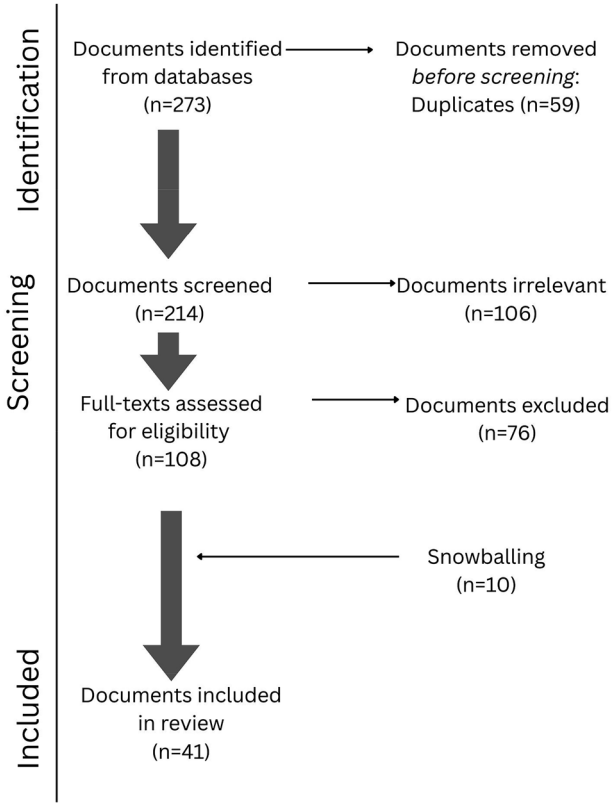

Fig. 1 Document filtering process.

Demographic reporting varied widely across studies (with some not reporting any demographic information), and few claimed to include representative or demographically diverse samples. Groups notably underrepresented included men, people of non-European descent, individuals without tertiary education, and those from lower socioeconomic backgrounds [10, 17, 18, 25, 41]. Several studies acknowledged the potential for selection bias, noting that participants were often more interested in health research or more knowledgeable about NBS and genomics than the general population [10, 17, 22, 29]. This bias may have skewed findings toward more polarised attitudes. Selection bias may have been especially pronounced in studies linked to pilot implementations of gNBS (such as the BabySeq study), as those studies were likely to have selected parents with a particular view. Efforts to mitigate selection bias and bias from prior knowledge were

Table 1. Overview of included documents and protocols.

Country	Year published	Title	Author	Associated Pilot Study	Participant group	Method	Themes
Australia	2024	Australian Public Perspectives on Genomic Newborn Screening: Risks, Benefits, and Preferences for Implementation	Lynch et al.	BabyScreen+	public (including those with a genetic condition in the family)	focus groups	acceptability, consent, psychosocial implications, data storage and use
Australia	2024	Australian public perspectives on genomic newborn screening: which conditions should be included?	Lynch et al.	BabyScreen+	public (including those with a genetic condition in the family)	focus groups	screening scope, equity and accessibility
Australia	2022	Australian healthcare professionals' perspectives on the ethical and practical issues associated with genomic newborn screening	Cao et al.	BabyScreen+	healthcare professionals (with genetics and/or NBS experience)	interviews	acceptability, consent, screening scope, psychosocial implications, data storage and use, healthcare readiness, equity and accessibility
Australia	2023	Are We Ready for Whole Population Genomic Sequencing of Asymptomatic Newborns?	Vears et al.	BabyScreen+	bioethicists, researchers	symposium	acceptability, consent, screening scope, psychosocial implications, data storage and use, healthcare readiness
Australia	2023	Expanding the Australian Newborn Blood Spot Screening Program using genomic sequencing: do we want it and are we ready?	White et al.		parents (including those with a genetic condition in the family), healthcare professionals (with genetics and/or NBS experience)	surveys	acceptability, consent, screening scope, psychosocial implications, data storage and use, healthcare readiness, equity and accessibility
Australia	2024	Key informant perspectives on implementing genomic newborn screening: a qualitative study guided by the Action, Actor, Context, Target, Time framework	Tutty et al.	BabyScreen+	healthcare professionals (with genetics and/or NBS experience), patient group members	interviews	acceptability, consent, psychosocial implications, healthcare readiness
UK	2021	Implications of whole genome sequencing for newborn screening: A public dialogue	Hopkins et al.	The Generation Study (formerly the Newborn Genomes Programme)	public (including those with a genetic condition in the family)	public dialogue	acceptability, consent, screening scope, psychosocial implications, data storage and use, healthcare readiness, equity and accessibility
UK	2022	Black Mums Supporting Babies Workshop Report	The Motherhood Group	The Generation Study	parents	workshop	acceptability, consent, psychosocial implications, equity and accessibility
UK	2023	Newborn Genomes Programme: a dialogue with ethnic minority community sector organisations	Genomics England	The Generation Study	public	interviews and workshops	acceptability, consent, psychosocial implications, data storage and use, equity and accessibility
UK	2023	Newborn Genomes Programme: Pilot Public Standing Group on Ethics	Kinsella et al.	The Generation Study	public	workshops	consent, psychosocial implications, data storage and use, healthcare readiness

Table 1. continued

Country	Year published	Title	Author	Associated Pilot Study	Participant group	Method	Themes
UK	2022	Newborn Genomes Programme: Establishing principles for feeding back results to families	Involve	The Generation Study	public (including those with a genetic condition in the family), healthcare professionals, patient group members	workshops, survey, interviews	consent, screening scope, psychosocial implications, healthcare readiness, equity and accessibility
UK	2022	A Public Dialogue to Inform the Use of Wider Genomic Testing When Used as Part of Newborn Screening to Identify Cystic Fibrosis	Kinsella et al.		public	longitudinal public dialogue	screening scope
UK	2021	YouGov Results - Genetic Testing	YouGov		public	survey	acceptability
UK	2024	Views of children and young adults about Whole Genome Sequencing in newborn screening: a qualitative study	Parfett et al.		public, patients (with a genetic condition)	focus groups, diaries	acceptability, consent, screening scope, psychosocial implications
UK	2024	Perceptions of genomic newborn screening: a cross-sectional survey conducted with UK medical students	Seed et al.		medical students	survey	acceptability, screening scope, psychosocial implications, data storage and use, healthcare readiness, equity and accessibility
Canada	2014	Public views on participating in newborn screening using genome sequencing	Bombard et al.		public	survey	acceptability
Canada	2023	Parental Preferences for Expanded Newborn Screening: What Are the Limits?	Liang et al.		parents	survey	acceptability, consent, screening scope, psychosocial implications
USA	2015	Parents are interested in newborn genomic testing during the early postpartum period	Waisbren et al.	BabySeq	parents (including those with a genetic condition in the family)	survey	acceptability
USA	2016	Psychosocial Factors Influencing Parental Interest in Genomic Sequencing of Newborns	Waisbren et al.	BabySeq	parents (including those with a genetic condition in the family)	survey	acceptability
USA	2019	Parental interest in genomic sequencing of newborns: enrolment experience from the BabySeq Project	Genetti et al.	BabySeq	parents	survey	acceptability, psychosocial implications, data storage and use
USA	2019	Perceived Benefits, Risks, and Utility of Newborn Genomic Sequencing in the BabySeq Project	Pereira et al.	BabySeq	parents, healthcare professionals (predominantly without genetics training)	survey	acceptability, consent, screening scope, psychosocial implications, data storage and use
USA	2021	Psychosocial Effect of Newborn Genomic Sequencing on Families in the	Pereira et al.	BabySeq	parents	survey	screening scope, psychosocial implications

Table 1. continued

Country	Year published	Title	Author	Associated Pilot Study	Participant group	Method	Themes
		BabySeq Project: A Randomised Clinical Trial					
USA	2023	Parents' decision-making regarding whether to receive adult-onset only genetic findings for their children: Findings from the BabySeq Project	Pereira et al.	BabySeq	parents	interviews	acceptability, consent, screening scope, psychosocial implications, data storage and use
USA	2022	Parental Attitudes Toward Standard Newborn Screening and Newborn Genomic Sequencing: Findings From the BabySeq Study	Armstrong et al.	BabySeq	parents	survey	acceptability, consent, screening scope
USA	2021	Effects of participation in a U.S. trial of newborn genomic sequencing on parents at risk for depression	Schwartz et al.	BabySeq	parents	suvrey, interviews	psychosocial implications
USA	2023	Perspectives of Rare Disease Experts on Newborn Genome Sequencing	Gold et al.	BabySeq	healthcare professionals and scientists with expertise in rare disease	survey	acceptability, screening scope
USA	2024	Genetic counsellors' perspectives on genomic screening of apparently healthy newborns in the United States	del Rosario et al.	BabySeq	healthcare professionals (with genetics experience)	survey	acceptability, screening scope, psychosocial implications, data storage and use, healthcare readiness, equity and accessibility
USA	2022	Diverse Parental Perspectives of the Social and Educational Needs for Expanding Newborn Screening through Genomic Sequencing	Timmins et al.	GUARDIAN	parents (including those with a genetic condition in the family)	interviews	acceptability, consent, screening scope, psychosocial implications, data storage and use
USA	2021	A behaviour-theoretic evaluation of values clarification on parental beliefs and intentions toward genomic sequencing for newborns.	Paquin et al.	NC Nexus	parents (including those with a genetic condition in the family)	experiment with decision aids	consent
USA	2020	Values clarification and parental decision making about newborn genomic sequencing	Peinado et al.	NC Nexus	parents (including those with a genetic condition in the family)	experiment with decision aids	consent
USA	2022	Parental Guidance Suggested: Engaging Parents as Partners in Research Studies of Genomic Screening for a Paediatric Population	Powell et al.	NC Nexus	parents	Community Research Board meetings	consent, screening scope, data storage and use, equity and accessibility
USA	2022		Goldenberg et al.		NBS officials	focus groups	acceptability, consent, screening scope, data

Table 1. continued

Country	Year published	Title	Author	Associated Pilot Study	Participant group	Method	Themes
		Genomics and Newborn Screening: Perspectives of Public Health Programs					storage and use, healthcare readiness, equity and accessibility
USA	2014	Parents' interest in whole-genome sequencing of newborns	Goldenberg et al.		parents	survey	acceptability, data storage and use
USA	2016	Parental Views on Expanded Newborn Screening Using Whole-Genome Sequencing	Joseph et al.		parents (including those with a genetic condition in the family)	focus groups	acceptability, consent, screening scope, psychosocial implications, data storage and use
USA	2015	Genetics professionals' opinions of whole-genome sequencing in the newborn period	Ulm et al.		healthcare professionals (with genetics experience)	survey	acceptability, consent, screening scope, data storage and use, healthcare readiness
USA, Canada	2024	Genomic counselling in the newborn period: experiences and views of genetic counsellors	Nardini et al.		healthcare professionals (with genetics experience)	survey	consent, screening scope, healthcare readiness
USA	2024	Charting the Ethical Frontier in Newborn Screening Research: Insights from the NBSTRN ELSI Researcher Needs Survey	Unnikumaran et al.		researchers, NBS officials, healthcare professionals, patient group members, parents (including those with a genetic condition in the family), patients	survey	acceptability, consent, data storage and use
NZ	2016	Parents' experiences 12 years after newborn screening for genetic susceptibility to type 1 diabetes and their attitudes to whole-genome sequencing in newborns	Kerruish et al.		parents (including those with a genetic condition in the family)	interviews	acceptability, screening scope, psychosocial implications
Slovenia	2023	Perception of genomic newborn screening among peripartum mothers	Prosenc et al.		parents (including those with a genetic condition in the family and experience with genetic testing)	survey	acceptability, screening scope, psychosocial implications, data storage and use
Bulgaria	2017	Whole-Genome Sequencing in Newborn Screening - Attitudes and Opinions of Bulgarian Paediatricians and Geneticists	Iskrov et al.		healthcare professionals (including those with genetics and/or NBS experience)	survey	acceptability, screening scope, psychosocial implications, data storage and use, healthcare readiness
China	2022	Are We Ready for Newborn Genetic Screening? A Cross-Sectional Survey of Healthcare Professionals in Southeast China	Wu et al.		healthcare professionals (with NBS experience)	survey	acceptability, screening scope, psychosocial implications, data storage and use
Protocols							
Australia	2024	Genetic newborn screening stakeholder perspectives	Kariyawasam et al.		parents, healthcare professionals, researchers, policymakers	interviews	

Table 1. continued

Country	Year published	Title	Author	Associated Pilot Study	Participant group	Method	Themes
Australia	2024	Prospective cohort study of genomic newborn screening: BabyScreen+ pilot study protocol	Lunke et al.	BabyScreen+	parents, public, healthcare professionals	focus groups, interviews, discrete choice experiements	
UK	2024	Exploring the feasibility, acceptability and impact of genomic newborn screening for rare diseases in England: A study protocol for the Generation Study - Process and Impact Evaluation	Lewis et al.	The Generation Study	parents, healthcare professionals, rare disease community, public, researchers, industry, policymakers	surveys, interviews	
UK	2024	The Generation Study Protocol	Tuff-Lacey et al.	The Generation Study	parents, healthcare professionals, rare disease community, public	meetings, interviews, coproduction, focus groups, presentations and deliberative workshops	
USA	2022	Implementation of Whole Genome Sequencing as Screening in a Diverse Cohort of Healthy Infants	Green et al.	BabySeq2	parents	surveys	

described in a minority of studies (e.g., by limiting information in recruitment materials [19]).

Healthcare professionals were included in several studies, spanning a range of roles and levels of genetic expertise. Samples included, clinical geneticists [11, 16, 34, 36, 41], genetic counsellors [34, 41, 45], and a range of representatives from mainstream specialities such as nurses [34], paediatricians [16], neonatologists [18], midwives [41], primary care providers [34], and medical students [46]. The extent to which specific healthcare professionals were engaged was often unclear as they were typically recruited as part of broader healthcare professional samples and their perspectives were not reported separately. Reporting on participants' prior experience with newborn screening was also limited.

This lack of differentiation is notable given the distinct roles these professionals are likely to play in practice. For instance, paediatricians and midwives—who are likely to play key roles in the delivery and communication of gNBS—were only rarely the focus of dedicated analysis. Only one study explicitly compared paediatricians with genetic specialists, finding that paediatricians were generally more supportive of genomic sequencing in newborn screening, whereas genetic specialists more frequently emphasised concerns related to uncertainty, counselling requirements, and the right not to know [16].

While the inclusion of a range of professional perspectives suggest some diversity of perspectives, there remains limited work with non-genetics specialists who will play an important role in mainstreaming gNBS. Recruitment often occurred through professional networks, and demographic details of participants were rarely reported. As with the other stakeholder groups, selection bias may have been at play in the recruitment meaning that there was a greater likelihood of sampling people with more polarised views than the broader workforce [34, 36, 45].

Finally, some studies included experts, such as bioethicists, laboratory scientists, policy advisors, and newborn screening program officials [26, 40, 47]. Notably, no study explicitly included health economists or related financial experts. These participants were relatively underrepresented compared with other stake holders.

How are gNBS issues being constructed?

Eligible studies were conducted across eight countries with the majority being from Anglophone countries (the USA, Australia, and the UK). All studies were institution led, representing perspectives that were prompted and gathered by research groups, although a few used co-creation methods or advisory panels to shape their work (i.e. patient and public engagement to inform research methods). Over half (24/41) of studies were linked to an offer of genomic newborn sequencing through a scientific research pilot, namely BabyScreen+ [34, 40, 42, 43, 48, 53], BabySeq [12, 15, 17, 18, 22, 23, 25, 36, 38, 45, 49], the Generation Study [20, 27, 29, 31, 35, 50, 52], and the NC Nexus study [19, 21, 30].

Various data collection methods were used to gather stakeholder perspectives, including interviews [23, 27, 32, 34, 35, 38, 48, 51, 52], focus groups [13, 26, 42–44, 50, 53], surveys [9–12, 15–18, 22–25, 27, 33, 36, 37, 39, 41, 45–47, 49, 52], diaries [44], and discussion groups via public dialogues, workshops, and symposia [20, 27–31, 35, 50]. A minority of studies used some form of longitudinal data collection through the organisation of multiple data collection points rather than a single event [28–30]. Broadly, more frequently used methods such as surveys tended to offer more limited opportunities for engagement, whereas interactive or iterative approaches (e.g. workshops, dialogues, longitudinal designs) allowed for more in-depth exploration, although this varied depending on study design.

How was information communicated to participants?. As gNBS is both novel and complex, nearly all studies provided some

Fig. 2 Systemic Mapping. Cells indicate the number of studies using a given method with a given stakeholder group. Darker shading indicates a greater number of studies. Studies engaging multiple stakeholder groups and/or using multiple methods are counted in more than one cell.

background information to participants. The methods used to prepare participants for engagement with gNBS varied substantially across studies, ranging from minimal background information to multi-session structured education. It was difficult to fully assess the content provided to participants as many studies only provided limited information in their description of the study methods. Several reported only that participants were given brief explanations of gNBS, with little detail on the content provided. In some cases, no preparatory materials were reported at all. In those that provided more explanation, background was often provided in the form of written descriptions, flowcharts, consent model outlines, or structured welcome packs. Vignettes describing categories of conditions (e.g., treatable childhood-onset, untreatable, adult-onset) were also used to anchor discussion. Several studies used short educational videos. For example, participants were shown 3-min videos explaining DNA, genomic sequencing, and current NBS procedures in Australia [42, 43]. Public dialogues and workshops frequently included presentations from experts including clinicians, scientists, and ethicists (such as those conducted by Genomics England [20, 27, 28]). Several of these studies also had discussion moderators, although it varied whether these were methodological or content experts. From dialogue and workshop reports, it was unclear the degree to which they intervened, if at all, when participants exhibited misunderstandings or misconceptions. For example, in the public dialogue organised by Genomics England [20], several participants expressed perspectives that appeared to be underpinned by a belief in genetic determinism. A smaller number of studies included one-on-one or group sessions with genetic counsellors to explain risks, benefits, and implications of participation [17, 18]. In community-based research, investigators provided historical context on past abuses in genetics research, followed by topical presentations and group discussions, fostering trust and shared understanding over multiple sessions [30]. With the exception of a minority of studies that incorporated explicit and structured approaches to directly assess comprehension (such as quizzes) [9], few studies explicitly evaluated the effectiveness of their strategies of communicating about gNBS. While some methods (such as diaries, interviews, or focus groups) may have provided opportunities to explore participants' understanding, this was rarely described or assessed in a systematic or explicit manner, leaving it unclear how well participants understood the complexities of the topic before offering their perspectives.

Topic coverage. Across included studies, seven main themes emerged regarding perceived benefits, challenges, and ethical considerations of gNBS: acceptability ($n = 31$), consent ($n = 24$), scope of screening ($n = 26$), psychosocial implications ($n = 25$), data storage and use ($n = 22$), healthcare readiness ($n = 13$), and equity and accessibility ($n = 11$) (Fig. 3).

Most studies examined stakeholder interest and motivation to participate in gNBS and framed their inquiries in terms of the perceived benefits and challenges. Most approached these issues through the lens of parental decision-making for newborns, leaving largely unexplored the perspective of individuals who would have been the recipients of genomic newborn screening. Decision-making and consent content often focused on consent models and the information required for meaningful consent. Scope of screening discussions frequently explored questions of treatability and actionability, as well as the additional uncertainty that genomic technologies could introduce into existing NBS. However, few studies explicitly addressed predictive limitations of screening and genomic information such as false positives and negatives, incomplete penetrance, or variable expressivity.

Psychosocial implications ($n = 25$) were another common focus, with studies exploring parental anxiety, the impact of uncertain or unexpected results, and broader family and social consequences of genetic information. Data storage and use ($n = 22$) were

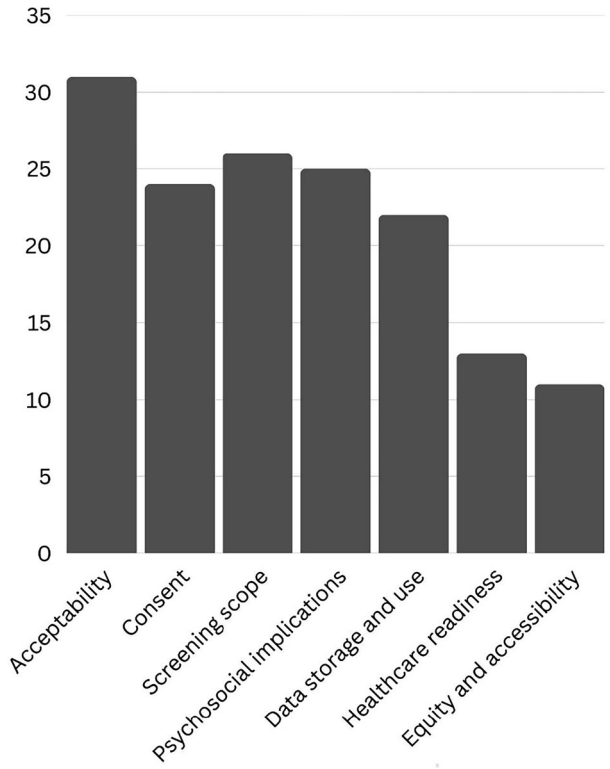

Fig. 3 Topic coverage.

recurrent themes, though discussions tended to emphasise risks of discrimination, data misuse, and unauthorised access by insurers, employers, or commercial entities, rather than the long-term implications of maintaining and potentially reinterpreting genomic data over the life course.

Healthcare readiness ($n = 13$) and accessibility ($n = 11$) were less frequently examined. When raised, health system concerns often centred on cost-effectiveness, workforce capacity, and the need for training and infrastructure to support implementation. Discussions of equity and accessibility were comparatively rare, predominantly covered by countries with publicly funded healthcare systems despite their salience for all public health programs. Coverage of accessibility issues were relatively superficial, typically with only mention of potential disparities.

DISCUSSION

This review aimed to critically examine how and in whom stakeholder perspectives on gNBS have been investigated. By evaluating which perspectives remain underrepresented and methods of data collection, we hope to contextualise the literature to make recommendations regarding interpretation and future research priorities.

We identified several gaps in stakeholder perspectives reported in the research on gNBS. Assessment of these gaps was complicated by inconsistent reporting practices of participant demographics. This conflicts with recent critiques on genomics research, which emphasised the need for transparent reporting of demographic data to promote accountability and diversity in participation [54]. From what could be discerned, participants were predominantly female, white, highly educated, and already had experience with genomics. These findings echo prior work investigating the landscape of genomics engagement demonstrating how certain populations remain underserved by research in genomics [55]. To address these gaps in representation, several groups tried to engage and recruit from underrepresented populations, either by engaging with pre-existing community groups (such as Genomics England's discussions with the Motherhood group, a support group for Black mothers [31]) or by recruiting participants of diverse backgrounds to form a community research group to advise researchers (such as in the NC Nexus Study [30]). Despite these initiatives and targeted recruitment efforts, participation remained low indicating that more work is needed to make research on gNBS better reflect the population that gNBS would eventually serve if implemented. Furthermore, special attention is needed to ensure that when members of underrepresented communities do participate, that their perspectives do not remain marginalised due to persistent power dynamics [56]. Ensuring the inclusion of underrepresented populations is critical not just in principle, but also in how it contributes to which ethical issues arise and are given weight in discourse on gNBS. Our review found that topics such as equity and access were only covered by a minority of studies. Likewise, few studies acknowledged how the lack of diversity in genomic reference data may affect the accuracy and relevance of gNBS results and downstream care.

Several studies, including those described as engaging the general public, drew heavily from rare disease communities. This is understandable, as these groups have strong motivations to participate. However, these are not the only perspectives relevant to the discourse on population screening of (apparently) healthy newborns where the ratio of true and false positive results will be very different. As gNBS moves toward possible population-wide implementation, future studies must engage participants with no prior personal or familial connection to genomics or rare disease to ensure findings reflect a diverse range of perspectives and experiences.

Healthcare professionals were another key stakeholder group, yet most participants were specialists from genetic services. Perspectives from midwives, paediatricians, and general practitioners remain underexplored despite the central role that they will likely have in delivering gNBS. Similarly, few studies included public health experts or screening programme directors, and no study included health economists, despite their expertise in population-level screening and resource allocation.

The limited experiences and perspectives represented by the work done on gNBS thus far may be particularly problematic in research linked or embedded within gNBS research pilots. In several studies, participants were recruited directly from a pool of parents that were interested in receiving gNBS through the research pilot. Even when this was not the case, if participants were aware that there was a gNBS pilot, or plans to have one, they may have been less critical or even inclined to give viewpoints that support gNBS. Studies embedded within implementation pilots may be constrained in their critical scope. One can imagine that it might be difficult for an ethics work package attached to a sequencing pilot to conclude that gNBS should not move forward without conflicting with the broader programme objectives. Independent research efforts will therefore be crucial to ensure a full range of perspectives are included in gNBS discourse.

Our analysis also highlights how study design and framing shape the views that are ultimately captured. A challenge of gathering perspectives on topics as complex and novel as gNBS is that it requires information provision for participants. Studies varied widely in how they prepared participants, from rich multimedia briefings to minimal information provision. The content and tone of this preparatory material or engagement can influence responses. Framing gNBS through cases of preventable tragedy (e.g. a child who could have been saved) will surely elicit different reactions than cases highlighting false positives (e.g. a child who could be harmed), uncertain results, or psychosocial burdens. Striking a balance between enabling informed engagement and avoiding undue framing effects remains a persistent methodological tension. Rather than

attempting to avoid biasing participants by only offering limited information, another potentially useful approach could be to expose participants to different types of framing to see how the connotation of the provided information impacts the formation of their perspectives [28, 44]. For example, in a longitudinal, multi-session format, participants could first be invited to share initial reactions based on minimal information, before being sequentially exposed to different framings of gNBS across subsequent sessions. These might include perspectives emphasising potential benefits (such as early detection and treatment for rare conditions), potential risks (such as false positives, uncertain findings, or the creation of 'patients-in-waiting'), and broader public health considerations (such as resource allocation and population-level trade-offs). Such an approach allows for the exploration of how participants' views evolve in response to contrasting information and would make the influence of framing an object of analysis rather than something to be avoided. It is critical that researchers practice reflexivity and acknowledge how even the act of asking certain questions inevitably signals which issues are deemed important. While fully unprompted perspectives are difficult to capture in the context of an emerging or future service such as gNBS, researchers could incorporate approaches that reduce the degree of researcher direction. For example, observational components could be embedded within pilot programmes to examine how information about gNBS is communicated and discussed in practice. In addition, methods such as digital ethnography or other minimally structured approaches could be used to capture how individuals encounter, interpret, and discuss gNBS in their everyday lives.

Broadly speaking, studies focused on trying to answer the question of whether or not stakeholders would support gNBS. In this, they explored several ethical issues, but the one that was reported on in the most depth was "informed consent". This is perhaps understandable, as consent has often been seen as a pillar of biomedical ethics and is relatively tractable for empirical study. Asking participants to choose between consent models is more straightforward than answering questions around what degree of uncertainty is acceptable. Informed consent alone cannot address the broader ethical challenges posed by gNBS [57], as obtaining informed consent will not ensure ethical implementation. Concerns such as equity and accessibility remain largely underexplored, despite their importance for gNBS if it is to transition from the research setting to implementation in public health systems.

Participant responses frequently reflected a broader societal tendency toward the assumption that genomic information offers clear, definitive predictions about health outcomes. In many studies, participants expressed confidence that genomic results would provide actionable information about their child's future health. Most genomic findings, particularly in the screening context, involve probabilistic risk estimates, mediated by complex interactions. Yet few studies actively worked to correct the baseline of genetic determinism that appeared to shape some perspectives, and in some cases, the explanatory study materials may have inadvertently reinforced misconceptions about the predictive value of genomics. These assumptions should be taken into account when interpreting attitudinal findings, and future engagement efforts will need to more clearly situate gNBS within the realities of genomic uncertainty [18, 28, 39, 42, 44]. Furthermore, when interpreting findings, it is important to acknowledge how findings may have biases, particularly given that more than half of the studies included in this analysis were embedded within a project with a gNBS pilot study. If participants were aware that a pilot study was planned or was occurring simultaneously, it could impact responses.

Finally, we observed variation in how findings were interpreted and presented. Some drew strong conclusions about the acceptability or desirability of gNBS based on small, non-representative samples. Others translated complex, ambivalent qualitative data into simplified claims of public support. These reductive summaries risk marginalising "inconvenient complexities" [58] and, when cited uncritically, may create a cumulative narrative of consensus that is not substantiated by the underlying evidence. While most studies openly acknowledged their limitations, these caveats often disappeared in secondary citation or policy translation, where findings are taken as evidence of public enthusiasm for gNBS. For instance, a recently published scoping review of public and parent perspectives on genomic newborn screening [8], found that these stakeholders are broadly accepting or have positive attitudes towards gNBS. While such reviews provide valuable overviews of reported attitudes, our findings suggest that these conclusions should be interpreted with caution. Across the studies included in our mapping, expressions of interest or support were frequently conditional, context-dependent, or accompanied by significant concerns regarding uncertainty, psychosocial impact, and broader societal implications. Furthermore, our analysis indicates that this positivity may, at least in part, be shaped by recruitment and study design, and may potentially be a reflection a wider issue in public discourse and communication around scientific and medical advances [55, 59]. As the empirical literature increasingly informs decision-making, it is vital that researchers and policymakers resist the temptation to conflate limited or qualified support with population-level endorsement.

CONCLUSION

As gNBS moves from research to potential public health implementation, understanding *whose* perspectives have shaped the evidence base and *how* these perspectives have been sampled is crucial. Our review reveals that while the existing literature is expanding rapidly, it remains uneven in whose voices are represented and which ethical issues are prioritised. Without addressing these gaps, there is a real risk that future gNBS policies will reflect a narrow subset of experiences and perspectives.

As ongoing gNBS pilots generate empirical data, it will be essential to capture participants' lived experiences—not only of positive findings, but also of negative and false positive results, and of navigating follow-up testing and care. These insights, especially the long-term psychosocial and practical impacts, will take time to emerge. In the meantime, it is vital to interpret current research findings with a critical lens, acknowledging the limitations that shape what we know so far.

High rates of support for gNBS in surveys and public dialogues should not be mistaken for a mandate to proceed with implementation. Public enthusiasm often reflects hopes for early diagnosis and better outcomes [60], but it may also be influenced by genetic determinism and incomplete understandings of what genomic information can and cannot deliver. These findings should not be taken as direct policy prescriptions, but as insights into expectations, concerns, and values that can guide how gNBS is communicated and implemented [10, 43]. Professionals delivering gNBS may need support in managing public expectations about the predictive limits of genomic data and the uncertainties it can introduce.

For policymakers, this review underscores the importance of interpreting stakeholder research not as a referendum on whether gNBS should proceed, but as a reflection of how it might be governed ethically and equitably. For researchers, it highlights the need to think critically about the purpose of engagement: is it just to decide whether gNBS should go ahead, or is it to deepen understanding of how people make sense of genomic information, navigate uncertainty, and weigh the social and ethical implications of screening? We as a community must be careful not to reduce diverse, nuanced perspectives into mere rhetorical support for competing sides of wider political and scientific debates. Decisions about implementation involve complex technical, ethical, and governance considerations that extend

beyond any single stakeholder group. Engagement should not aim to adjudicate policy decisions, but rather to deepen understanding of how people experience the ethical and social issues raised by genomic technologies. Decisions about implementation ultimately rest with public health authorities, but those decisions must be grounded in methodologically rigorous and ethically reflective research that captures a full diversity of experiences, perspectives, and values. Providing this foundation is a central responsibility of the research community.

DATA AVAILABILITY

The data that support the findings of this study are available upon request from the corresponding author.

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AUTHOR CONTRIBUTIONS

Conceptualisation: AP, KL, AML, CFW, LJ. Literature search: DK, AP. Formal analysis: AP, LJ. Funding acquisition: AML, CFW, LJ. Project administration: AP, LJ, CFW. Visualisation: AP. Writing-original draft: AP. Writing-review & editing: AP, KL, DK, AML, CFW, LJ.

FUNDING

This research was supported by the MRC [MR/X021351/1]. AP, CFW, and LJ were also supported by the National Institute for Health and Care Research Exeter Biomedical Research Centre. The views expressed are those of the authors and not necessarily those of the NIHR or the Department of Health and Social Care.

COMPETING INTERESTS

The authors declare no competing interests.

ETHICAL APPROVAL

Ethical approval was not required for the conduct of this study as this was a mapping of existing literature.

ADDITIONAL INFORMATION

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